Mind the Gap

See related article, p 94

Before 1972, pediatricians generally relinquished the care of teenage patients to adult providers. That year, a forward-thinking American Academy of Pediatrics (AAP) Section on Child Health redefined pediatric care to encompass birth through age 21 years. Dedication to longitudinal care for teens and young adults was sealed through creation of certification in Adolescent Medicine through the American Board of Pediatrics in 1991. Decades later, it is clear that our AAP and American Board of Pediatrics leaders were prescient in this approach. Each subsequent decade brings increasing numbers of youth with chronic disease and medical complexity into adulthood. Recent insight into the impact of social determinants and adverse childhood events on longitudinal health has increased provider attention on the unequivocal impact of psychosocial variables on patient health. The mandate for this vigilance is not limited to primary and preventive care but also includes subspecialists managing pediatric chronic disease. The manuscript by Michel et al in this volume of *The Journal* further illuminates this challenge while revealing gaps in care among a subset of such youth and teens, ie, those living with inflammatory bowel disease (IBD).[1]

IBD is a group of chronic systemic diseases including both Crohn's disease and ulcerative colitis. Nearly 1.5 million Americans are affected, with ~25% presenting before 20 years of age. Although IBD often is considered in the family of childhood-onset chronic disease, unlike type 1 diabetes mellitus, sickle cell disease, cystic fibrosis, and others, the most common age of onset for pediatric IBD is during adolescence and not early childhood. This feature of the illness affects numerous aspects of patient care (eg, issues of autonomy, body image, and growth) and likely impacts patient and family readiness for transition to adult care.

In the study by Michel et al, parents of children aged 2-17 years and teens themselves (aged 13-17 years) were recruited from one academic center's IBD clinic.[1] They were surveyed regarding aspects of primary and pediatric gastroenterology-focused care they recall receiving. Although all youth had seen their gastroenterology provider in the past 12 months, 88% also had seen their primary care provider (PCP), in most cases for well preventive care. This is significantly greater than in broad US data sets, which put the number of youth seeing a PCP in the same age group and time frame as just less than one-half.[2,3] In the survey group, 94% were of white race, 80% privately insured, and almost all parents were college educated. This suggests that the study population may have been less racially diverse and more affluent than is typical for pediatric IBD. The methodology and single site of data collection is inherently burdened by both potential recall and ascertainment bias. Regardless, the authors reveal critical gaps in psychosocial care that are impactful and generalizable for providers and recipients of pediatric primary and specialty care nationwide. They open doors for collaborative opportunities to enhance child health, specifically for adolescents.

Current AAP Bright Futures Recommendations and Guidelines for Adolescent Preventive Services rightly pair immunizations, physical growth, and development with the now-imperative screening and counseling around social determinants of health, emotional well-being, risk reduction, and safety practices.[4] PCPs are aided by validated screening tools, notably the HEADSS (home, education and employment, activities, drugs [including tobacco and alcohol], sex, suicidality/depression) mnemonic for history-taking, the CRAFFT (CAR, RELAX, ALONE, FORGET, FRIENDS, TROUBLE) screen for identifying substance abuse risks, and the PHQ-9 (Patient Health Questionnaire) screen for early detection of depression and suicidality.[4] Yet outpatient primary care visits rarely allow sufficient time for these. In the data set of Michel et al, the largest gaps in recollected care involved discussion around mood, sexuality, and transition to adult care. From these gaps, opportunities emerge.[1]

Depressive symptomatology and its most feared outcome, suicidal ideation, continue to increase among older teens and young adults, keeping suicide as the second-leading cause of death among Americans aged 15-34 years. Several papers document an increased prevalence of depression and an almost 8-fold incidence of suicidality among those with IBD.[5,6] The PHQ-9, a simple self-administered tool, is well validated and indispensable for rapid identification of those at risk within a primary care setting. Lister et al validated the use of this tool to detect suicidal ideation among a large US and Canadian population with IBD.[6] Implementation of some mental health screening is feasible in subspecialty clinics. In 1 study, depression screens were administered in gastroenterology and endocrine clinics for patients with IBD, diabetes mellitus, or cystic fibrosis. A 15% positive screening rate for depression was found in this group across clinics.[7] It is time to add this approach to the subspecialist's toolkit for IBD and other chronic illnesses.

Although adolescent pregnancy has decreased over the last decade, likely due to widespread messaging around condom use and receipt of long-acting reversible contraception, the

AAP American Academy of Pediatrics
IBD Inflammatory bowel disease
PCP Primary care provider

M.C. is an Editorial Board member of *The Journal of Pediatrics*. The authors declare no conflicts of interest.

https://doi.org/10.1016/j.jpeds.2020.05.054

imperative for healthy discussion on this topic remains. By age 16 years, the probability of sexual debut among current US teens remains around 35% and increases to almost 75% by age 19 years.[8] More than one-half of youth have viewed Internet pornography by age 11 years, with a 93% view rate by age 18 years.[8] The late teen years present a critical time for provider discussion and demystification around factual sexuality and reproductive health. The critical yet challenging basis of such conversations is confidentiality. Several surveys show adolescents' willingness to openly talk to physicians about substance use, mood, and sexuality when they are given alone time and assured of privacy.[9] The AAP has long upheld such confidentiality within adolescent visits. Despite this, less than one-half of adolescents report time alone with clinicians during preventive visits,[9] and it is likely even fewer have time alone when visiting their gastroenterologist. This is a missed opportunity. Among teens and parents, familiarity with providers is the tenet most important to accepting and engaging in confidential communication. Telemedicine has been used to improve rates of IBD follow-up care.[10] With the recent dramatic increase in use of telemedicine, this may be another successful way to engage in these confidential conversations in a non–office-based environment for some adolescents.

The final critical gap emerging from this study is discussion around transition to an adult IBD provider. Although transition as a process for youth with chronic disease has been studied exhaustively, determinants of successful transition remain elusive. Qualitative analysis of patients with IBD, parents, and providers brings insight that confirms the outlook of many primary care pediatricians. Patients overwhelmingly desire transition to adult care at a time of both life and disease stability.[11,12] They want a definitive time, a "red line in the sand." Many youth view completion of college, military service, or full-time workforce entry as a tangible and preferred "red line," choosing then to ascend the first step in transition to ownership of their self-care. Transition to an adult provider makes more sense to patients when they know where they are going to settle and/or work after graduation; it makes less sense to transition twice. In part because of this, with good merit, the AAP saw reason to extend the upper limit of pediatric care to age 22 years.

From these gaps come opportunities. Pediatric IBD physicians are pediatricians first and understand the magnitude of psychosocial modifiers on longitudinal disease management and health. Patients and families often revere their continuity with their pediatric IBD specialty teams, particularly through the tumultuous period of adolescence. Because at most one-half of adolescents receive preventive primary care, specialist initiative and teamwork are critical. Discussions involving emotional health and risk taking are an opportunity for specialists and should be part of pre-visit planning. They are often blessed with patient trust, and unlike many PCPs, they have extended visit time and co-located nutritionists, educators, social workers, and behavioral health providers. Shared expectations around timing for and concrete aspects of disease ownership between family, adolescent youth, PCP, and specialty providers is critical. Having adolescents make their own appointments, check in, request refills, know medication doses, intervals and side effects, and their baseline laboratory values, if applicable, is part of the expectation of transition and preparation for self-care. With the advent of linked electronic medical records, encrypted e-mail servers, and the knowledge that real-time communication is embraced by PCPs, shared screening begets more communication. When one provider initiates, the other can reinforce. The bottom line for today's teens is that adolescence portends great vulnerability. Transition requires time and communication. Although this is not a one-size-fits-all process, all adolescents must transition. As the authors state, coordinated, comprehensive care delivery models are needed for this important life and care event. ■

Morissa J. Ladinsky, MD
Mitchell B. Cohen, MD
University of Alabama at Birmingham
Birmingham, Alabama

Reprint requests: Mitchell B. Cohen, MD, Children's of Alabama, 1600 7th Ave South, Birmingham, AL 35233. E-mail: mcohen@peds.uab.edu

References

1. Michel HK, Kim SC, Siripon N, Noll RB. Gaps exist in the comprehensive care of children with inflammatory bowel diseases. J Pediatr 2020;224:94-101.
2. Rand C, Goldstein N. Patterns of primary care physician visits for US adolescents in 2014: implications for vaccination. Acad Pediatr 2018;18: S72-8.
3. Adams SH. Association between adolescent preventive care and the role of the Affordable Care Act. JAMA Pediatr 2018;172:43-8.
4. Hagan JF, Shaw JS, Duncan PM, eds. Bright Futures: guidelines for health supervision of infants, children, and adolescents. 4th ed. Elk Grove Village (IL): American Academy of Pediatrics; 2017.
5. Fuller-Thomson E, Sulman J. Depression and inflammatory bowel disease: findings from two nationally representative Canadian surveys. Inflamm Bowel Dis 2006;12:697-707.
6. Lister B, Bernstein C, Graff L. Validation of the PHQ-9 for suicidal ideation in persons with inflammatory bowel disease. Inflamm Bowel Dis 2018;24:1641-8.
7. Iturralde E, Adams RN, Barley RC, Bensen R, Christofferson M, Hanes SJ, et al. Implementation of depression screening and global health assessment in pediatric subspecialty clinics. J Adolesc Health 2017;61:591-8.
8. Abma I, Martinez G. Sexual activity and contraceptive use among teenagers in the USA, 2011-2015. Natl Health Stat Report 2017;104:1-23.
9. Miller V, Friedrich E. Adolescents spending time alone with pediatricians during routine visits: perspectives of parents in a primary care clinic. J Adolesc Health 2018;63:280-5.
10. Dykes D, Williams E, Margolis P, Ruschman J, Bick J, Saeed S, et al. Improving pediatric Inflammatory Bowel Disease (IBD) follow-up. BMJ Qual Improv Rep 2016;5. u208961.w3675.

11. Gray W, Resmini A, Baker KD, Holbrook E, Morgan PJ, Ryan J, et al. Concerns, barriers, and recommendations to improve transition from pediatric to adult IBD care: perspectives of patients, parents, and health professionals. Inflamm Bowel Dis 2015;21:1641-51.

12. White PH, Cooley WC. Transitions Clinical Report Authoring Group; American Academy of Pediatrics; American Academy of Family Physicians; American College of Physicians. Supporting the health care transition from adolescence to adulthood in the medical home. Pediatrics 2018;142:e20182587.

The Harms of Carrier Status Identification: A Cautionary Warning Against Newborn Sequencing

Two articles by Farrell et al in this volume of *The Journal* explore the potential psychosocial complications of carrier status notification for sickle cell hemoglobinopathy (SCH) and cystic fibrosis (CF) after newborn screening (NBS). Both articles stem from the Wisconsin Project on Improvement of Communication and Process Outcomes after Newborn Screening. In the first, qualitative telephone interviews with parents were conducted after NBS carrier status was disclosed by a primary care provider to evaluate for effectiveness of results communication, misconception of the child's risk for disease, and parental anxiety.[1] The second assessed parents' perceptions of child vulnerability after being informed of carrier result for SCH or CF, and both groups were compared with a control group using an adapted version of the Vulnerable Baby Scale.[2] Notably, the data collected by Farrell et al in both studies was gathered between 2008 and 2012, and the findings reported in these 2 articles have been corroborated in other studies for these same diseases: (1) parental misunderstanding of carrier status for children with SCH or CF, even in a state that offers genetic counseling; (2) parental anxiety or stress from receiving incidental information; and (3) increased parental assessment of child vulnerability after carrier identification.[3-6] And yet, despite reaffirming the potential harms of carrier status identification in NBS, Farrell et al expect it to expand, concluding that they "suspect that genome sequencing on blood spots will be routine within the coming generation, regardless of ELSI [ethical, legal, and social implications] concerns." Below, we explore why the data from Farrell et al further strengthen the ethical, legal, and social concerns and reject the inevitability of universal adoption of genomic sequencing into NBS programs.

See related articles, p 37 and p 44

NBS has traditionally focused on conditions and disorders, like CF and SCH, that present early in infancy for which early diagnosis can prevent morbidity or mortality. Although most screening currently uses tandem mass spectrometry, the appeal of whole genome sequencing is the potential to screen and diagnose even more conditions using a single platform. However, genomic sequencing without phenotypic information still misses many cases of conditions that are currently identified in NBS. In 2014, Bhattacharjee et al attempted to identify the conditions included in state NBS panels.[7] They wrote: "It is typically assumed that, at least for monogenic disorders, the genotype-phenotype relationship would be simple." Instead the authors found their "ability to pinpoint the clinical phenotype of an individual on the basis of 'genotype' alone is still in its infancy; in our case, only 27 of 36 NBS disease cases were classified correctly without phenotype information."[7]

But imagine that sequencing was better able to identify the conditions included in state NBS panels and could be implemented as the primary platform for NBS. Screening for more conditions would also mean identifying many more carriers. Although broad professional consensus in the US in the early 1990s led to the decision to disclose carrier status when identified in NBS, all US professional statements argue against routine carrier identification in children.[8-11] In BabySeq, a study exploring genomic sequencing of both infants in the neonatal intensive care unit and healthy infants, the researchers demonstrated that >90% of infants screened had ≥1 carrier status variant, with an average of 2 carrier status variants and a range from 0 to 7.[12] This finding is lower than data from Bell et al, who found the average participant (noninfant) on whom genomic sequencing was performed was a carrier for 2.8 conditions (range, 0-7).[13] Primary care physicians are already ill-equipped to discuss NBS carrier results with parents.[14,15] The identification of more carrier status variants in infants through sequencing will only exacerbate these issues; more information about newborns is not always better, particularly when the information is nonactionable for the health of the infant. Farrell et al have demonstrated the possibility of harm to parents and their children from returning these ancillary results—particularly to parents of lower health literacy.[1,2] The failure to effectively counsel a significant number of parents about carrier status

CF	Cystic fibrosis
IRT	Immunoreactive trypsinogen
NBS	Newborn screening
SCH	Sickle cell hemoglobinopathy

L.R. is an Editorial Board member for *The Journal of Pediatrics*. The authors declare no conflicts of interest.

https://doi.org/10.1016/j.jpeds.2020.05.014